The role of the physiotherapist in Huntington’s disease develops in response to the clinical need over the prolonged course of the illness; it is important for physiotherapists to consider the cost implications of their services throughout a lifelong disease.

Physiotherapy management should be tailored to the stage of the disease, but also importantly to specific signs and symptoms that affect functional performance and life participation; treatment-based classifications can be used to better categorize patient problems and guide specific physiotherapy interventions.

Physiotherapy management should begin soon after diagnosis, with a focus on encouraging an active lifestyle, and maximizing and maintaining physical functioning.

Early referral enables practitioners to determine a baseline for the person with Huntington’s disease, and supports the establishment of a therapeutic relationship between the person with Huntington’s disease, practitioners and caregivers.

During early and middle stages of the disease, physiotherapy evaluation should include measures to aid in clinical decision making, including identification of common impairments, such as bradykinesia, dystonia and postural imbalances.

Therapists should consider use of responsive clinical measures to aid in outcomes assessment and falls risk assessment, utilizing tools such as the Berg Balance Scale, Tinetti Mobility Scale, Timed Up and Go Test and measures of gait speed.

Towards the middle to end stages, therapists should work in collaboration with other healthcare professionals to achieve the best possible quality of life for patients and their families; therapists have an important role in advising on seating and positioning and respiratory management, as well as relaxation techniques.
SUMMARY  

Background: Physiotherapy may provide a means of delaying onset or progression of Huntington’s disease (HD), resulting in improved daily functioning and quality of life. Physiotherapy is being more frequently recommended for people with HD, but there have been no specific guidelines published for implementation of a structured physiotherapy program. The Physiotherapy Working Group (PWG) of the European Huntington’s Disease Network (EHDN) set out to develop a comprehensive Guidance Document for physiotherapists to provide best practice guidelines. Methods: A review of the literature was conducted using a systematic approach. There was insufficient literature in support of physiotherapy interventions and approaches to be able to conduct a complete evidenced-based review, therefore, physiotherapy expert subgroups were formed to incorporate consensus as to best practice. A draft document was distributed to the entire membership of the working group, to outside physiotherapists and other healthcare professionals within EHDN to elicit feedback and comments. Results: A Guidance Document covering eight specific areas pertaining to physiotherapy management of HD was developed. In order to facilitate the document’s practical usability among clinicians, a treatment-based classification system is proposed to categorize patients based on presenting signs and symptoms, and provide a foundation for development of a more standardized intervention approach. Discussion: The Physiotherapy Guidance for HD is a comprehensive, consensus- and evidence-based document that can be used by physiotherapists to implement a plan of care that is currently consistent with best practice for individuals at all stages of HD. As evidence becomes available, future systematic reviews will be required in order to inform further development. The use of treatment-based classifications, which aim to better categorize common signs and symptoms and link them to appropriate intervention plans, may be useful in relatively rare diseases, such as HD, to aid clinical reasoning and promote effective outcome evaluation.

Huntington’s disease (HD) is a degenerative neurological disease that results in a triad of clinical symptoms, encompassing motor, behavioral and cognitive impairments. Researchers have developed a better understanding of the complex nature of the movement disorder in this disease, however, to date, physiotherapy intervention is not provided consistently [1]. The beneficial role of physiotherapy for people with Parkinson’s disease, a basal ganglia disorder with some similar features to HD, has been previously illustrated, with two recent systematic reviews reporting that physiotherapy can improve multiple factors, including physical functioning, health-related quality of life (HR-QoL), strength, balance and gait [2,3]. The literature in support of physiotherapy for people with HD is less clear. Two reviews have noted that to date there has been only a small amount of evidence in support of physiotherapy within HD [4,5]. Although the impact of currently available literature is somewhat overshadowed by poor methodological rigor, small sample sizes, unclear selection criteria (resulting in potential heterogeneity in participant groups) and a lack of follow-up, there have more recently been some optimistic developments. For example, a before–after trial with a sample size of 40 found an intensive rehabilitation program of six sessions per week demonstrated an improvement in motor function over the 2-year period [6,7]. Positive findings from environmental enrichment studies in mice also provide some support for the role of physiotherapy for HD. In mice models of HD, those mice placed within an environment providing physical, mental and social stimulation, have a slower disease progression, and maintain motor function for longer compared with those housed in more isolated conditions [8,9].

People with HD have a range of physical, cognitive, psychological and social care needs over an extended time frame. One of the difficulties in developing clinical guidelines for complex neurodegenerative diseases, such as HD, is the heterogeneity of clinical signs and symptoms. While staging of the disease process (e.g., early, middle, late) can provide a general framework for intervention, within each stage there is a wide range of potential impairments that can impact on an individuals’ level of functional activity and life participation. This makes structuring of consistent therapeutic approaches problematic. This problem is not unique to HD and has been documented in other physiotherapy patient groups, most notably low back and neck dysfunction, and for general neurorehabilitation patient groups [10–15].
Researchers have advocated that by creating sub-classifications of patients’ impairments and problems, and matching those with more specific interventions, therapists may improve outcomes in their patients [10–12,15]. These researchers have argued that in order for therapists to develop consistent approaches to common patient problems, so that the efficacy of any one approach can be tested, better categorization as described above is required. Opinions differ as to whether this categorization should come in the form of a movement-based system (i.e., based on specific movement problems only), or one which assesses a wider range of signs and symptoms. Regardless, the resultant categorization can provide the foundation for development of a more standardized intervention approach.

Clinical guidelines are evidence-based recommendations for clinical practice in specific conditions [16,17]. The availability of clinical guidelines facilitates uniformity of care and standards of practice with the aim of improving quality of care provision. Availability of appropriate guidelines can change the process and practice of care for the benefit of the patient and can inform the research process, leading to more effective research into practice.

It is critically important for health professionals to be able to define and document their evaluation practices and intervention strategies, so that systematic outcome evaluation can be done and best practice highlighted. Clinical guidelines are in place for physiotherapy in other neurodegenerative diseases, such as Parkinson’s disease [13,18] and multiple sclerosis [19]. While physiotherapy is increasingly being proposed as a mode of intervention for people with HD, to date there has been no formalized clinical guidelines or standards of care to guide practice. The Physiotherapy Working Group (PWG) of the European Huntington’s Disease Network (EHDN), therefore, set out to develop a guidance document to provide a written framework for the physiotherapeutic management of people with HD. The aims were to provide, where possible, a scientific evidence-based document to inform the optimal, individualized physiotherapeutic management of people with HD. This included the provision of a written framework for the physiotherapeutic management of people with HD, in order to enable uniformity of care internationally. We also aimed to highlight the scientific evidence for physiotherapy practice with people with HD, evaluating the effectiveness of any intervention strategies presented in the literature. A guidance document would allow practitioners to make an informed decision regarding patient care and to highlight areas within practice that currently lack the scientific evidence base to inform future research.

Methods

The Physiotherapy Guidance Document for HD was created through a combination of available scientific evidence and expert consensus. A PICO (Population, Intervention, Comparison, and Outcome) approach to searching the literature was used. The population was defined as people with HD. The intervention was any physiotherapy-based intervention or assessment. No set comparisons were made or follow-up times set. Outcomes were any reliable and valid measure of physiotherapy-related interventions.

Databases searched were Ovid MEDLINE® 1950 to March Week 2 2009; EMBASE 1980 to 2009 (Week 12, 21 March); EBSCO CINAHL 1981 to 21 March 2009; PEDro 1929 to 21 March 2009. Search terms included: Huntington’s disease, chorea, Huntington, Huntington chorea; physical therapy modalities, physiotherapy, rehabilitation, occupational therapy, physical activity, exercise therapy, activities of daily living/activities of daily living, physical mobility, muscle stretching exercises/stretching, respiratory therapy/respiratory therapy, flexibility, range of motion, range of movement, upper extremity/upper limb, reaching, reach, grasping, grasp, reach to grasp, posture, balance, accidental falls/falls, equipment and supplies/equipment, seating, positioning, transfers, functional status, gait.

Inclusion criteria were: articles published in English; therapy in human HD subjects only; subjects to be of 18 years of age and with a confirmed diagnosis of HD; study specific to physiotherapy/occupational therapy/rehabilitation and exercise. A total of 239 individual papers were identified through database searching. A total of 206 of these were excluded, leaving a total of 33 papers for review. The main reasons for exclusion were for non-human studies, subjects did not have HD, and the study was not related to physiotherapy/occupational therapy/rehabilitation or exercise. This left a total of 31 papers for review in the full guidance document. Two of these were not retrieved and 33 were summarized in the full guidance document.
Three reviews, nine physiotherapy-related studies, 11 gait-specific, one muscle strength, three balance and mobility, one dystonia, two upper limb-specific and one paper regarding multisensory stimulation were identified. The identified evidence was summarized independently and then discussed at an EHDN PWG meeting. This summary is presented in the final guidance document. Owing to the paucity of scientific evidence, recommendations were also formulated based on expert consensus from the EHDN PWG. Following completion of the initial draft document, the Guidance Document was disseminated to all members of the PWG and other interested healthcare professionals. Amendments were made to the document based on their feedback. In a second phase of development, sections of the Guidance Document were reviewed in detail by subgroups of two to three members of the PWG. Feedback from the subgroups were directed back to the authors, and integrated into the final document.

Following publication and dissemination of the Guidance Document, the PWG met and discussed the impact of the document and provided constructive feedback. A consensus was reached that the next step in development of the PWG was to provide a user-friendly format that made it readily useful to novice therapists, but one that would not be limiting or overly prescriptive for more experienced clinicians. The methodology provided by treatment-based classification systems was an obvious choice for structuring such guidance. The group agreed that current research in the area of treatment-based classification [10–12,15] had potential for application to the HD population. We therefore proposed a range of treatment-based classifications to address the heterogeneity of patient conditions, to provide structure for standardized data collection of interventions and outcomes, and consequently inform evaluation of complex interventions and advance research into care and evidence-based service delivery for people with HD. A second subgroup was subsequently formed, consisting of expert physiotherapists within HD and neurodegenerative diseases. Two focus group meetings of this group were held over a 6-week period, during which time treatment-based classifications were constructed using the Guidance Document as a template for discussions. An iterative process was undertaken where key impairments were classified, so that the key signs and symptoms commonly found in HD could be organized within a classification system. The discussions also focused on linking these classifications with specific interventions and outcome measures from the Guidance Document.

Results

Physiotherapy guidance for HD
The review of the literature revealed a lack of any well-controlled randomized intervention studies of physiotherapy interventions in HD that could be used to conduct a systematic review. We considered all relevant literature and case reports, as well as observational studies that were identified in the review to provide some insight as to the nature of the impairments that could provide a theoretical foundation and recommendations for physiotherapy interventions. Alongside this process of reviewing the literature, we consulted with expert clinicians to finalize our recommendations.

A brief overview of the philosophy and recommendations for developing physiotherapy plans as well as considerations of procedural interventions is presented. Here we report the summary recommendations. Greater detail on each topic as well as the review of the literature can be found in the formal physiotherapy guidance document [101].

Framework for patient management
An important component to patient management in a neurodegenerative disease such as HD is to consider people at all stages of the condition, including those who have the mutation for HD but are not displaying motor symptoms (pre-manifest) [1,20]. It is further suggested that physiotherapy management of people with HD should be modified according to individual problems and to the stage of the disease. The reader is referred to the original paper where the framework development is described [1]. Currently, intervention focuses on symptomatic management, however, there is increasing support for early intervention where an impact may be made on biological processes with the potential to influence the natural history of the condition. In addition, early referral to physiotherapy for people with
HD may be beneficial in a number of ways. It enables practitioners to ascertain a baseline for the person with HD, supports the establishment of a therapeutic relationship between the person with HD, practitioners and caregivers, and ensures early intervention to attempt to maintain mobility and function for as long as possible [1].

Evaluation & outcome measures
The physiotherapy evaluation for people with HD should be both anticipatory and responsive to the disease stage. The physiotherapist should work in collaboration with the patient and evaluate their individual considerations [21]. The WHO International Classification of Functioning, Disability and Health (ICF) [22] can aid assessment of a person’s level of functioning, and further facilitate consideration of the triad of motor, cognitive and psychiatric symptoms that are often seen in HD. It can also be used for goal setting and treatment planning, and focuses on aspects of a person’s health and health-related wellbeing in terms of activities and participation, that is, the description of the tasks (activities) and/or life situations (participation) the person wishes to be involved in, and the impact that impaired body function or structure is having on these aspects.

A structured evaluation for the patient with HD is comprised of both subjective and objective assessments, the primary goal being to determine the relationship between any impairments, activity limitations and participation restrictions. Caregiver involvement, wherever possible, is strongly encouraged, particularly if the patient has difficulties communicating through cognitive or physical impairment. The objective evaluation should include assessment of neuromuscular (e.g., bradykinesia, dystonia, chorea), musculoskeletal (e.g., posture, range of motion, pain, muscle strength) and cardiorespiratory and cardiovascular impairments (e.g., vital signs, breathing function, exercise capacity). Standardized tests can be utilized as outcome measures that may be sensitive to changes related to any physiotherapy intervention. The Berg Balance Scale, Tinetti Mobility Scale, Timed Up and Go Test, Four square step test and the Functional Reach Test have all been found to be valid and responsive clinical measures in HD and are considered useful to detect those at risk of falls [23–26].

Physiotherapy interventions
Since pharmacologic interventions to date have been ineffective in altering or slowing the disease process, healthcare practitioners and patients themselves continue to look for alternative therapies. While physiotherapy has been shown to be of benefit for some neurodegenerative diseases, such as Parkinson’s disease and multiple sclerosis, the evidence supporting its use for people with HD is limited. Nonetheless, utilizing traditional physiotherapy techniques with applicability to the special needs of this population may provide some benefit for a disease that currently lacks any effective treatment regimes.

The main goals of physiotherapy intervention will generally change over time. In the early stages, interventions will be primarily preventative. As the disease progresses, interventions become restorative in nature, addressing specific physical impairments and resulting activity limitations. In the later stages, intervention is primarily compensatory, in which therapists develop strategies to maximize a patient’s functional ability despite progression of the disease. With regards to falls risk, a clear justification of benefit of any particular management strategy compared to the risk that the situation presents should be elucidated. Therapists working with people with HD should acknowledge and facilitate independent mobility for as long as is reasonably possible [1].

Goals should be specific and measurable and mutually agreeable to both the patient and therapist [27]. They should address specific functional problems or participation restrictions that are amenable to physiotherapy intervention. Goals to decrease chorea or dystonia, for example, may not be realistic. Furthermore, amelioration of any particular impairment in HD may not translate into functional improvements and, therefore, therapists should focus on functional gains while attempting to ascertain the influence various impairments have on activity limitations in each of their patients.

Similar to healthy individuals, exercise, including strengthening and cardiovascular conditioning, is recommended for people who are pre-manifest and in the early stages of the disease. Exercise may be helpful in minimizing any neuromuscular or musculoskeletal effects that come with disease progression. As the disease progresses and specific problems become evident, therapists must begin to ascertain those impairments that may be contributing to specific
activity limitations or participation restrictions. For example, involuntary movements could contribute to balance problems, but inactivity may also be a contributing factor. Impairment-based diagnoses that may impact on function include: dystonia; bradykinesia; moderate-to-severe chorea or ballismus; rigidity; impaired respiratory function and fatigue. It is important to manage the secondary effects of such impairments. For example, for patients with chorea, protective equipment can be provided, and for patients with dystonia, loss of range of motion and muscle imbalance should be prevented. Functional problems that may occur include: impaired fine motor skills and manual dexterity, impaired sitting posture and sitting ability, impaired mobility, transfers and gait; impaired balance/and risk of falls and reduced cardiovascular and general physical fitness [1,23–25]. In general, it is thought that physical fitness plays a part in a patient’s motivation to keep as active as possible and their ability to cope with more challenging treatment interventions.

The potential impact of psychiatric impairments and a patient’s cognitive status cannot be overlooked. Therapists should consider whether or not a patient has memory loss, depression, aggression, obsessive–compulsive tendencies or anxiety, to name a few, and these impairments must be taken into account during the design of any intervention plan [28].

Documentation & interdisciplinary management
Developing and documenting a plan of care for the patient with HD should also include coordination of services and communication that needs to take place with other professionals, caregivers or family members so that the physiotherapy plan can be implemented. This may also include referrals to other professionals and additional documentation of any planned home program and patient/caregiver education. Importantly, the physiotherapist should be part of an interdisciplinary team that is involved in the management of patients with HD throughout the lifespan of their disease. Such professionals include geneticists, neurologists, nurses, occupational therapists, psychologists, speech language therapists and social workers, among others. It is critical for therapists to consider collaboration with these professionals and to understand their role as part of an interdisciplinary team and in particular their contribution to palliative care in the late stages of the disease. The ultimate aim for palliative care is to achieve the best possible quality of life for patients and their families [29]. The physiotherapist can advise on seating and positioning, respiratory management as well as relaxation techniques and may also have a role to play when considering advanced care directives [30].

Cost implications
It is also important for physiotherapists to consider the cost implications of their services throughout a life-long disease. It is unreasonable to expect that any funding agency (government or otherwise) would be agreeable to ongoing physiotherapy intervention two- to three-times a week for the extended disease process. Instead, therapists should consider serving consultative roles to patients from an early stage, and providing more intensive intervention when a change in functional status warrants it.

Treatment-based classifications
Seven treatment-based classifications specific to HD have been developed using the Guidance Document as a reference point. The primary findings discussed above for the Guidance Document were utilized to categorize patients more specifically based on primary impairments and activity limitations (Table 1). Within each classification, we determined signs and symptoms as a method for categorization. We then listed general aims, and provided specific intervention strategies and outcome measures. Examples of classifications are available from the authors, and will be made available in full on the EHDN website in due course. Clinical validation of these classifications is currently underway across the EHDN network.
healthcare professional information days across the UK and Europe.

**Discussion**

While physiotherapy is increasingly being considered an important mode of intervention for people with HD, to date there has been no formalized guidance or standards of care to document best practice. A need was identified from within the HD community to first and foremost conduct pragmatic analysis of current physiotherapy service delivery in HD, and to document the available evidence related to physiotherapy services. This paper describes the process and outcomes from the development of these clinical guidelines for physiotherapy management of people with HD.

Owing to the progressive nature of HD, the needs of people with this condition change over time, and the role of the physiotherapist should be flexible in response to the clinical need over the prolonged course of the illness. Although the evidence supporting physiotherapy interventions for people with HD is limited, there is some suggestion from HD literature and from other similar neurodegenerative diseases that exercise and other physiotherapy strategies may be useful in minimizing the effects of this devastating disease. The Physiotherapy Guidance Document has been developed based on expert consensus and available evidence, and is freely available to the HD community. It does, however, continue to be a work in progress and will require review and updating as new more robust research becomes available.

The availability of a guidance document is the first step towards standardization of physiotherapy practice. We must however highlight that the background research done to support the guidance document yielded a paucity of applicable research studies, and no randomized controlled trials aimed at evaluating the efficacy of physiotherapy or exercise. The challenge of evaluating physiotherapy is largely due to problems of identifying, documenting and reproducing the multiple components of the intervention. The randomized controlled trial was originally developed for evaluation of single interventions, such as a particular drug, and is not fully compatible with complex interventions, such as physiotherapy. It is clear that rigorous evaluation of complex interventions such as physiotherapy is imperative to their continued existence, but this requires a phased approach that can ultimately lead to a definitive controlled trial [31]. The Medical Research Council Framework for evaluation of complex interventions refers to a theoretical and a modeling stage prior to conducting full-scale randomized controlled clinical studies [31]. The first step is to review the relevant theory and literature that is available to ensure best design of intervention; this is called the theoretical phase. The next modeling phase involves identifying components of the intervention of choice (based on the previous theoretical stage) and determining how the outcomes would be

<table>
<thead>
<tr>
<th>Classification</th>
<th>Description of clinical findings</th>
<th>Stages of disease</th>
</tr>
</thead>
<tbody>
<tr>
<td>A. Exercise capacity and</td>
<td>Absence of motor impairment; potential for cognitive and/or behavioral issues</td>
<td>Presymptomatic/early</td>
</tr>
<tr>
<td>performance</td>
<td></td>
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<tr>
<td>B. Planning and sequencing of</td>
<td>Difficulty and slowness in performing functional activities (e.g., dressing, bathing, ADLs or sit to stand). Presence of apraxia or impaired motor planning; slowness of movement and/or altered force generation capacity</td>
<td>Early-mid</td>
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<td>tasks (including bradykinesia)</td>
<td></td>
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<tr>
<td>C. Mobility, balance and</td>
<td>Ambulatory for community and/or household distances; balance, strength or fatigue resulting in falls or high risk for falls</td>
<td>Early-mid</td>
</tr>
<tr>
<td>falls risk</td>
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<td></td>
</tr>
<tr>
<td>D. Secondary adaptive changes</td>
<td>Musculoskeletal and/or respiratory changes resulting in decreased participation in daily activities</td>
<td>Early-mid</td>
</tr>
<tr>
<td>and deconditioning</td>
<td></td>
<td></td>
</tr>
<tr>
<td>E. Abnormal posturing (seating</td>
<td>Inappropriate alignment due to adaptive changes, involuntary movement, inability to facilitate or coordinate movement</td>
<td>Mid-late</td>
</tr>
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<td>and bed positioning; manual</td>
<td></td>
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<td>handling)</td>
<td></td>
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<td>F. Respiratory dysfunction</td>
<td>Impaired respiratory function and capacity; limited endurance; impaired airway clearance; risk for infection</td>
<td>Mid-late</td>
</tr>
<tr>
<td>G. Palliative care</td>
<td>Unable to ambulate; dependent for most ADLs; difficulty maintaining upright sitting position; range of motion and pulmonary issues</td>
<td>Late</td>
</tr>
</tbody>
</table>

**Table 1. Seven treatment-based classifications developed for physiotherapy management of people with Huntington’s disease.**

ADL: Activities of daily living.
achieved. It is clear that mixed methodology incorporating both quantitative and qualitative methods are relevant to these initial stages. The Guidance Document developed for HD used this phased approach and provides a starting point for the theoretical and modeling components of the framework. It provides the foundations for development of future physiotherapy-related controlled trials.

Development of treatment-based classifications as an extension of the Guidance Document can help to further facilitate standardization of practice. This approach requires an expansion and some modification of the conceptual framework presented [1], which bases intervention strategies on stages of the disease [1,20]. Treatment-based classifications address the issue of heterogeneity within HD by targeting specific problems that are seen irrespective of stage of disease. This allows for a more individualized approach to patient management but incorporates a clear structure for evaluation of efficacy.

Limitations, implications & future directions

The development of this Guidance Document and the subsequent classification system is the first step in the process towards solidifying the evidence base in physiotherapy in HD. The literature review for this document was limited to English-speaking articles, and thus should be a starting point for a more extensive multilanguage search. An important next step is clinical validation and revision of the classifications, in an effort to promote standardization of care. Once standardization of care is achieved, we can take the next important step in evaluating the efficacy of our interventions through pragmatic controlled clinical trials.

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Financial & competing interests disclosure

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No writing assistance was utilized in the production of this manuscript.

References

Papers of special note have been highlighted as:


2. Highlights physiotherapists’ views on provision of services for people with Huntington’s disease (HD) in the UK, and emphasizes the need for intervention in the early stages.


6. Highlights key aspects of motor impairments found in people with HD, and outlines a framework for physiotherapy evaluation and intervention in basal ganglia disorders.


8. Discusses caregivers’ perspectives on rehabilitation programs for HD.


12. Describes treatment-based classifications in general neuromuscular conditions.
Physiotherapy guidance & treatment-based classifications for people with HD

SPECIAL REPORT

Contributing members of the European Huntington’s Disease Network Physiotherapy Working Group: Guidance Document

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  - Sheila Watters
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Contributing members of the committee for the development of treatment-based classifications

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- Monica Busse
- Angela Hall
- Una Jones
- Hanan Khalil
- Lori Quinn

- Provides a framework for disease management in neurodegenerative diseases.
- Highlights barriers and facilitators to home- and community-based programs for people with HD and Parkinson’s disease.
- Describes useful clinical tools for measuring mobility and balance in HD.

Website